



OPHTHALMOLOGY PANELS

PANEL NAME	PHENOTYPES CONDITIONS	GENES
Achromatopsia panel		ATF6, CNGA3, CNGB3, CNNM4, GNAT2, OPN1LW, OPN1MW, PDE6C, PDE6H, RPGR
Albinism panel		AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1
Bardet-Biedl Syndrome panel		ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, IFT172, IFT27, LZTFL1, MKKS, MKS1, NPHP1, SDCCAG8, TRIM32, TTC8, TTC21B, WDPCP
Cataract		AGK, BCOR, BFSP1, BFSP2, CHMP4B, COL11A1, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTD1P1, EPHA2, EYA1, FOXC1, FOXE3, FTL, FYCO1, GALK1, GCNT2, GJA3, GJA8, HSF4, LIM2, LSS, MAF, MIP, NHS, OPA3,

		P3H2, PAX6, PITX3, PXDN, SIL1, SIPA1L3, SLC16A12, TDRD7, TFAP2A, TMEM70, UNC45B, VIM, VSX2, WFS1, WRN
Cone Rod Dystrophies panel		ABCA4, ACBD5, ADAM9, AIPL1, BEST1, C21orf2, C2orf71, C8orf37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP290, CERKL, CLN3, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, FBLN5, GNAT2, GUCA1A, GUCY2D, KCNV2, MERTK, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, TLL5
Congenital Stationary Night Blindness panel		CABP4, CACNA1F, CACNA2D4, GNAT1, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RBP4, RHO, SAG, SLC24A1, TRPM1, RDH5, RLBP1, RPE65, CACNA1, CABP4, CACNA1F, NYX
Familial Exudative Vitreoretinopathy and Wagner Syndrome panel		CAPN5, COL2A1, FZD4, LRP5, NDP, TSPAN12, VCAN, ZNF408
Flecked retina		CHM, EFEMP1, PLA2G5, RDH5, RLBP1, RS1, VPS13B
Glaucoma		ACVR1, ASB10, BEST1, CANT1, COL18A1, CYP1B1, FOXC1, LMX1B, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, PITX3, SBF2, WDR36, GLC3B, GLC3C, RIEG2, B3GALT1, NF1
Hereditary sensory autonomic neuropathy		ATL1, ATL3, DNMT1, DST, FAM134B, IKBKAP, KIF1A, NGF, SCN9A, SCN11A, SPTLC1, SPTLC2, RAB7, WNK1

Hermansky-Pudlak Syndrome		AP3B1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6
Joubert Syndrom panel		AHI1, ARL13B, B9D1, C5orf42, CC2D2A, CEP104, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIF7, NPHP1, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423
Leber Congenital Amaurosis panel		AIPL1, CABP4, CEP290, CRB1, CRX, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, NMNAT1, OTX2, PRPH2, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TULP1
Microphthalmia		ABCB6, ALDH1A3, BCOR, BMP4, CHD7, ERCC1, ERCC2, ERCC5, ERCC6, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GDF3, GDF6, GJA1, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SALL2, SHH, SIX3, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, VAX1, VSX2
Oculomotor apraxia		APTX, PIK3R5, PNKP, SETX
Ophthalmoplegia		DNA2, OPA1, POLG, POLG2, RRM2B, SLC25A4, TWNK, TYMP
Optic Atrophy panel		ACO2, AFG3L2, C12orf65, CISD2, MFN2, NR2F1, OPA1, OPA3, RTN4IP1, SLC25A46, SPG7, TIMM8A, TMEM126A, VAX2, WFS1

Refsum Disease panel		PEX1, PEX2, PEX26, PEX3, PEX5, PEX7, PHYH
Retinitis Pigmentosa, autosomal dominant and X-linked panel, 50 genes		ABCA4, ADGRA3, ARL3, BEST1, C2orf71, CA4, CERKL, CNGA1, CRB1, CRX, DHDDS, DHX38, FAM161A, FSCN2, GUCA1B, HK1, IDH3B, IMPDH1, IMPG2, KIAA1549, KLHL7, NR2E3, NRL, PDE6A, PDE6B, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP4, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPGR, SEMA4A, SNRNP200, SPATA7, SPP2, TOPORS, ZNF408
Retinitis Pigmentosa, autosomal recessive and X-linked panel, 70 genes		ABCA4, ADGRA3, ARL2BP, ARL3, ARL6, BBS1, BBS2, C2orf71, C8orf37, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CYP4V2, DHDDS, DHX38, EMC1, EYS, FAM161A, FLVCR1, FSCN2, GNPTG, GUCA1B, HGSNAT, HK1, IDH3B, IFT172, IMPG2, KIAA1549, KIZ, KLHL7, LRAT, MAK, MERTK, NEK2, NMNAT1, NR2E3, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPH2, RBP3, RBP4, RDH11, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SPATA7, SPP2, TOPORS, TRNT1, TTC8, TUB, TULP1, USH2A, ZNF408, ZNF513
Senior Loken Syndrome panel		CEP164, CEP290, IFT81, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19, ZNF423
Septo-optic Dysplasia	Optic nerve hypoplasia, Aniridia, Microphthalmia, Cataract with late-onset corneal dystrophy, Microphthalmia syndromic 3	LHX4, HESX1, POU1F1, PROP1, PAX6, SOX2, FGFR2, PROKR2, SOX3, TAX1BP3, OTX2
Stargardt Disease and Macular Dystrophies panel		ABCA4, BCA4, BEST1, C1QTNF5, CDH3, CNGB3, CLN3, CNGB3, CRB1, CRX, CTNNA1, DRAM2, ELOVL4, FSCN2, IMPG1, IMPG2, MFSD8, PRDM13,

		PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3, TTLL5
Stickler Syndrom panel		COL11A1, COL11A2, COL2A1, COL9A1, COL9A2
Usher Syndrome panel		ABHD12, ADGRV1, CDH23, CIB2, CLRN1, DFNB31, GPR98, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, USH1C, USH1G, USH2A

For more information contacts us: info@amplexa.com/ Ph+45 6611 6628